

ID	Chr	Mid point	Band	OMIM	Disease	Gene ID
RP1-283E3	1	1664815.5	1p36.33	607872	Monosomy 1p36	CDC2L2
RP1-140A9	1	1782951	1p36.33	607872	Monosomy 1p36	CDC2L2
RP11-120J4	2	15896442.5	2p24.3	164280	Feingold syndrome	MYCN
RP11-355H10	2	16031731	2p24.3	164280	Feingold syndrome	MYCN
RP11-495A22	2	16073403	2p24.3	164280	Feingold syndrome	MYCN
RP11-82K13	2	44856155.5	2p21	157170	Holoprosencephaly 2	SIX3
RP11-89K21	2	45008621.5	2p21	157170	Holoprosencephaly 2	SIX3
RP11-455L18	2	45021583.5	2p21	157170	Holoprosencephaly 2	SIX3
RP11-322F4	2	110153781	2q13	256100	Nephronophthisis 1/Joubert syndrome 4	NPHP1
RP11-528G9	2	110260893	2q13	256100	Nephronophthisis 1/Joubert syndrome 4	NPHP1
RP11-116D16	2	172519772	2q31.1	606708	Split-Hand/Foot Malformation 5	DLX1
RP11-472A3	2	172584628.5	2q31.1	606708	Split-Hand/Foot Malformation 5	DLX1
RP11-356J12	2	172710203	2q31.1	606708	Split-Hand/Foot Malformation 5	DLX1
RP11-514D19	2	176612729	2q31.1	186000	Synpolydactyly/Syndactyly Type II	HOXD13
RP11-387A1	2	176676919	2q31.1	186000	Synpolydactyly/Syndactyly Type II	HOXD13
RP11-437N19	2	176769512.5	2q31.1	186000	Synpolydactyly/Syndactyly Type II	HOXD13
RP11-497D24	2	238357849.5	2q37.3	600430	Brachydactyly-mental retardation syndrome	D2S2338
RP11-574K22	2	238548608.5	2q37.3	600430	Brachydactyly-mental retardation syndrome	D2S2338
RP11-526L8	2	238713348	2q37.3	600430	Brachydactyly-mental retardation syndrome	D2S2338
RP11-120J22	3	148539179.5	3q24	220200	Dandy-Walker syndrome	ZIC1 ZIC4
RP11-152O9	3	148612719	3q24	220200	Dandy-Walker syndrome	ZIC1 ZIC4
RP11-343P5	3	148671350	3q24	220200	Dandy-Walker syndrome	ZIC1 ZIC4
RP11-321C3	3	148761389	3q24	220200	Dandy-Walker syndrome	ZIC1 ZIC4
RP11-24F1	3	190732387.5	3q28	605289	Split-Hand/Foot Malformation 4	TP73L
RP11-364E11	3	190787473.5	3q28	605289	Split-Hand/Foot Malformation 4	TP73L
RP11-575K16	3	190969493	3q28	605289	Split-Hand/Foot Malformation 4	TP73L
RP11-373I6	3	191069928.5	3q28	605289	Split-Hand/Foot Malformation 4	TP73L
RP11-53D15	3	191167780.5	3q28	605289	Split-Hand/Foot Malformation 4	TP73L
RP11-200I19	3	197769046	3q29	609425	3q29 Microdeletion	PAK2
RP11-470E12	3	197928747.5	3q29	609425	3q29 Microdeletion	PAK2
RP11-296H14	3	198081581	3q29	609425	3q29 Microdeletion	PAK2
RP11-241P10	4	1775197.5	4p16.3	194190	Wolf-Hirschhorn syndrome	FGFR3 WHSC1
RP11-262P20	4	1946796.5	4p16.3	194190	Wolf-Hirschhorn syndrome	FGFR3 WHSC1
RP11-317B7	4	2058706	4p16.3	194190	Wolf-Hirschhorn syndrome	FGFR3 WHSC1
RP11-501E13	4	111492675.5	4q25	180500	Rieger syndrome Type 1	PITX2
RP11-380D23	4	111761127	4q25	180500	Rieger syndrome Type 1	PITX2
RP11-313B13	4	111837551	4q25	180500	Rieger syndrome Type 1	PITX2
RP11-554C16	5	9471938.5	5p15.31	123450	Cri-du-Chat syndrome	D5S630
RP11-32D12	5	9521103.5	5p15.31	123450	Cri-du-Chat syndrome	D5S630
RP11-340B04	5	9635832	5p15.31	123450	Cri-du-Chat syndrome	D5S630
RP11-513P01	5	9787754	5p15.31	123450	Cri-du-Chat syndrome	D5S630
RP11-91C21	5	36933941	5p13.2	122470	Cornelia de Lange syndrome	NIPBL
RP11-452E14	5	37040228.5	5p13.2	122470	Cornelia de Lange syndrome	NIPBL
RP11-7M4	5	37082803	5p13.2	122470	Cornelia de Lange syndrome	NIPBL
RP11-105B4	5	37130686.5	5p13.2	122470	Cornelia de Lange syndrome	NIPBL
RP11-56P2	5	112060770	5q22.2	175100	Adenomatous polyposis of the colon	APC
RP11-3B10	5	112077246.5	5q22.2	175100	Adenomatous polyposis of the colon	APC
RP11-107C15	5	112154847.5	5q22.2	175100	Adenomatous polyposis of the colon	APC
RP11-248F15	5	112265636.5	5q22.2	175100	Adenomatous polyposis of the colon	APC
RP11-160L22	5	176362941	5q35.2	130650	Beckwith-Wiedemann syndrome/Sotos syndrome	NSD1
RP11-116H24	5	176501263	5q35.2	130650	Beckwith-Wiedemann syndrome/Sotos syndrome	NSD1
RP11-99N22	5	176564901.5	5q35.2	130650	Beckwith-Wiedemann syndrome/Sotos syndrome	NSD1
RP11-265K23	5	176643005	5q35.3	130650	Beckwith-Wiedemann syndrome/Sotos syndrome	NSD1
RP11-394I17	6	45343999.5	6p21.1	119600	Cleidocranial dysplasia	RUNX2
RP3-427I13	6	45506667.5	6p21.1	119600	Cleidocranial dysplasia	RUNX2
RP11-342L7	6	45619725	6p21.1	119600	Cleidocranial dysplasia	RUNX2
RP1-166H4	6	45658836	6p21.1	119600	Cleidocranial dysplasia	RUNX2
RP11-113P17	6	45762126	6p21.1	119600	Cleidocranial dysplasia	RUNX2
RP11-117M4	6	100867444.5	6q16.3	603128	Prader-Willi-like phenotype	SIM1
RP1-121G13	6	101043443	6q16.3	603128	Prader-Willi-like phenotype	SIM1
RP11-296A15	6	101141465.5	6q16.3	603128	Prader-Willi-like phenotype	SIM1
RP11-139G22	7	41900988.5	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-282B02	7	41927573	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-762A04	7	42033712	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-472L24	7	42090969.5	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-259N24	7	42129866.5	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3

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RP11-325A19	7	42177737.5	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-96K1	7	42440980	7p14.1	175700	Greig cephalopolysyndactyly syndrome	GLI3
RP11-334D20	7	72336949	7q11.23	194050	Williams-Beuren syndrome	FZD9
RP11-101D2	7	72541227	7q11.23	194050	Williams-Beuren syndrome	FZD9
RP11-590H03	7	72743160.5	7q11.23	194050	Williams-Beuren syndrome	FZD9
RP11-73G23	7	72748160.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-148M21	7	72878654.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-644H24	7	73117706.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-100C23	7	73226583.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-331N10	7	73270547	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-422O01	7	73397615.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-329B05	7	73450792	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-247L6	7	73593297.5	7q11.23	194050	Williams-Beuren syndrome	ELN LIMK1 RFC2 CYLN2
RP11-474F4	7	96055545.5	7q21.3	183600	Split-Hand/Foot Malformation 1	SHFM1
RP11-95J17	7	96154943.5	7q21.3	183600	Split-Hand/Foot Malformation 1	SHFM1
RP11-94N7	7	96299670.5	7q21.3	183600	Split-Hand/Foot Malformation 1	SHFM1
RP11-120J15	7	155179759.5	7q36.3	142945	Holoprosencephaly 3	SHH
RP11-69O3	7	155271073	7q36.3	142945	Holoprosencephaly 3	SHH
RP11-451N23	7	155367421	7q36.3	142945	Holoprosencephaly 3	SHH
RP11-177H2	8	10715375	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-437P14	8	10932322.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-467A16	8	10989714.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-448A20	8	11188849	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-350A23	8	11271653	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-582P18	8	11343129	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-148O21	8	11456650.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-235I5	8	11630282.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-589N15	8	11802838	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-481A20	8	11854246	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-247B12	8	11899978.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-1236K1	8	12000014	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-354C4	8	12044729	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-351I21	8	12333948.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-419J17	8	12364200	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-303G3	8	12524643.5	8p23.1	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-813L8	8	12791244	8p22	147920	Kabuki syndrome 8p22 deletion/duplication syndrome	
RP11-91I20	8	61673911	8q12.1	214800	CHARGE syndrome	CHD7
RP11-414L17	8	61763761	8q12.1	214800	CHARGE syndrome	CHD7
RP11-331I1	8	61908419	8q12.2	214800	CHARGE syndrome	CHD7
RP11-359G10	8	72150283	8q13.3	113650	Branchiootorenal syndrome	EYA1
RP11-326E22	8	72213979.5	8q13.3	113650	Branchiootorenal syndrome	EYA1
RP11-11K9	8	72377419	8q13.3	113650	Branchiootorenal syndrome	EYA1
RP11-160C13	8	72392735	8q13.3	113650	Branchiootorenal syndrome	EYA1
RP11-10I24	8	116314649	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-104P5	8	116420683	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-466M24	8	116501409	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-762A3	8	116646303.5	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-164M9	8	116779107.5	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-66L24	8	116905573	8q23.3	190350	Trichorhinophalangeal syndrome Type I	TRPS1
RP11-494N20	8	118655548.5	8q24.11	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-111I15	8	118975962	8q24.11	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-260I14	8	119080033	8q24.11	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-346L16	8	119228241.5	8q24.11	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-455I9	8	119281603	8q24.11	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-103G12	8	119405237.5	8q24.12	150230	Trichorhinophalangeal syndrome Type 2	EXT1
RP11-346O18	9	97153352.5	9q22.32	109400	Basal cell nevus syndrome	PTCH
RP11-435O5	9	97268232	9q22.32	109400	Basal cell nevus syndrome	PTCH
RP11-205K6	9	128338499	9q33.3	161200	Nail-Patella syndrome	LMX1B
RP11-123K19	9	128441339.5	9q33.3	161200	Nail-Patella syndrome	LMX1B
RP11-489N22	9	128549381.5	9q33.3	161200	Nail-Patella syndrome	LMX1B
RP11-554F11	10	8079086	10p14	146255	Hypoparathyroidism sensorineural deafness and renal disease	GATA3
RP11-379F12	10	8160167.5	10p14	146255	Hypoparathyroidism sensorineural deafness and renal disease	GATA3
RP5-1119O21	10	8338983.5	10p14	146255	Hypoparathyroidism sensorineural deafness and renal disease	GATA3
RP11-348G8	10	12179978	10p14	601362	DiGeorge syndrome 2	D10S293
RP11-186N15	10	12302181.5	10p13	601362	DiGeorge syndrome 2	D10S293
RP11-477H7	10	12439311.5	10p13	601362	DiGeorge syndrome 2	D10S293
RP11-165K23	10	21034780	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-56H7	10	21144623	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-213K14	10	21303375	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-565H13	10	21581185.5	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-275N1	10	21705327.5	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-418C1	10	21853589	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-177H22	10	21955975	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-86F19	10	22045797	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-399C16	10	22151507.5	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-365J16	10	22304768	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-358M4	10	22386723	10p12.31	601362	DiGeorge syndrome 2	NEBL

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RP11-108B14	10	22487640.5	10p12.31	601362	DiGeorge syndrome 2	NEBL
RP11-264H19	10	103207502.5	10q24.32	600095	Split-Hand/Foot Malformation 3	SHFM3
RP11-148E14	10	103362073.5	10q24.32	600095	Split-Hand/Foot Malformation 3	SHFM3
RP11-534I22	11	1881563.5	11p15.5	130650	Beckwith-Wiedemann syndrome	H19 IGF2
RP3-416J11	11	2100843	11p15.5	130650	Beckwith-Wiedemann syndrome	H19 IGF2
RP11-889I17	11	2140420.5	11p15.5	130650	Beckwith-Wiedemann syndrome	H19 IGF2
RP5-1075F20	11	2294177	11p15.5	130650	Beckwith-Wiedemann syndrome	H19 IGF2
RP11-38L8	11	2715220	11p15.4	130650	Beckwith-Wiedemann syndrome	CDKN1C KCNQ1
RP11-774H09	11	2720189.5	11p15.4	130650	Beckwith-Wiedemann syndrome	CDKN1C KCNQ1
RP11-699D10	11	2924666.5	11p15.4	130650	Beckwith-Wiedemann syndrome	CDKN1C KCNQ1
RP11-304P12	11	3036306	11p15.4	130650	Beckwith-Wiedemann syndrome	CDKN1C KCNQ1
RP11-1146B5	11	3100249.5	11p15.4	130650	Beckwith-Wiedemann syndrome	CDKN1C KCNQ1
RP11-366N23	11	31682660.5	11p13	194072	WAGR syndrome	PAX6
RP11-133E13	11	31808608.5	11p13	194072	WAGR syndrome	PAX6
RP11-26B16	11	31830909.5	11p13	194072	WAGR syndrome	PAX6
RP4-760G15	11	32283116	11p13	607102	Wilms tumor 1/WAGR syndrome	WT1
RP1-74J1	11	32384348.5	11p13	607102	Wilms tumor 1/WAGR syndrome	WT1
RP1-259N9	11	32529632	11p13	607102	Wilms tumor 1/WAGR syndrome	WT1
RP11-70A9	11	43974289.5	11p11.2	601224	Potocki-Shaffer syndrome	ALX4 EXT2
RP11-224F21	11	44046539	11p11.2	601224	Potocki-Shaffer syndrome	ALX4 EXT2
RP11-104M24	11	44237945	11p11.2	601224	Potocki-Shaffer syndrome	ALX4 EXT2
RP11-70A24	11	44240252.5	11p11.2	601224	Potocki-Shaffer syndrome	ALX4 EXT2
RP11-450F4	11	44365506	11p11.2	601224	Potocki-Shaffer syndrome	ALX4 EXT2
RP11-715F10	11	67066561	11q13.2	161015	Leukodystrophy	NDUFV1
RP11-119D9	11	67364107	11q13.2	161015	Leukodystrophy	NDUFV1
RP3-521E19	12	111227208	12q24.13	163950	Noonan syndrome	PTPN11
RP3-329E11	12	111327536.5	12q24.13	163950	Noonan syndrome	PTPN11
RP1-66E7	12	111459839	12q24.13	163950	Noonan syndrome	PTPN11
RP5-1048I22	12	111539351.5	12q24.13	163950	Noonan syndrome	PTPN11
RP11-196P14	13	47683276.5	13q14.2	180200	Retinoblastoma	RB1
RP11-305D15	13	47829250.5	13q14.2	180200	Retinoblastoma	RB1
RP11-174I10	13	47879346	13q14.2	180200	Retinoblastoma	RB1
RP11-165D7	13	48038992.5	13q14.2	180200	Retinoblastoma	RB1
RP11-134O15	13	99304859.5	13q32.3	609637	Holoprosencephaly 5	ZIC2
RP11-12G12	13	99469390	13q32.3	609637	Holoprosencephaly 5	ZIC2
RP11-340C20	13	99620976.5	13q32.3	609637	Holoprosencephaly 5	ZIC2
RP11-385H1	15	22563293	15q11.2	176270	Prader Willi syndrome	SNRPN
RP11-125E1	15	22664541.5	15q11.2	176270	Prader Willi syndrome	SNRPN
RP11-98D02	15	22676506	15q11.2	176270	Prader Willi syndrome	SNRPN
RP11-642G3	15	22799900	15q11.2	176270	Prader-Willi syndrome	SNRPN
RP11-512F9	15	22811164	15q11.2	176270	Prader-Willi syndrome	SNRPN
RP11-99L13	15	22815426.5	15q11.2	176270	Prader-Willi syndrome	SNRPN
RP11-131I21	15	22815433.5	15q11.2	176270	Prader-Willi syndrome	SNRPN
RP11-720B15	15	23014322	15q11.2	176270	Prader-Willi syndrome	UBE3A
RP11-183K8	15	23026942.5	15q11.2	176270	Prader-Willi syndrome	UBE3A
RP11-234J13	15	23198807	15q11.2	105830	Angelman syndrome	UBE3A
RP11-466L14	15	23242570.5	15q11.2	105830	Angelman syndrome	UBE3A
RP11-173H16	15	23275532.5	15q11.2	105830	Angelman syndrome	UBE3A
RP11-208F21	15	23346688.5	15q12	105830	Angelman syndrome	UBE3A
RP11-52D3	15	91159780.5	15q26.1	142340	Diaphragmatic hernia 1 (congenital)	CHD2
RP11-437B10	15	91294557	15q26.1	142340	Diaphragmatic hernia 1 (congenital)	CHD2
RP11-368J22	15	91347501.5	15q26.1	142340	Diaphragmatic hernia 1 (congenital)	CHD2
RP11-302G10	15	94598258.5	15q26.2	142340	Diaphragmatic hernia 1 (congenital)	NR2F2
RP11-262A16	15	94612372	15q26.2	142340	Diaphragmatic hernia 1 (congenital)	NR2F2
RP11-522B15	15	94790161	15q26.2	142340	Diaphragmatic hernia 1 (congenital)	NR2F2
RP11-157H9	16	1850551	16p13.3	600273	Polycystic kidney disease/tuberous sclerosis 2	PKD1 TSC2
RP11-304L19	16	2167498.5	16p13.3	600273	Polycystic kidney disease/tuberous sclerosis 2	PKD1 TSC2
RP11-111H22	16	2429616	16p13.3	600273	Polycystic kidney disease/tuberous sclerosis 2	PKD1 TSC2
RP11-103P19	16	3630156	16p13.3	180849	Rubinstein-Taybi syndrome	CREBBP
RP11-75P12	16	3839145	16p13.3	180849	Rubinstein-Taybi syndrome	CREBBP
RP11-95J11	16	3882923.5	16p13.3	180849	Rubinstein-Taybi syndrome	CREBBP
RP11-294J05	17	1222125	17p13.3	247200	Miller-Dieker lissencephaly syndrome	YWHAE
RP11-210O04	17	1360822.5	17p13.3	247200	Miller-Dieker lissencephaly syndrome	YWHAE
RP1-59D14	17	2246558	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-98F18	17	2277688	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-135N5	17	2402099	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-582F10	17	2531046.5	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-74E22	17	2563355.5	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-102N8	17	2684005.5	17p13.3	247200	Miller-Dieker lissencephaly syndrome	LIS1
RP11-378M22	17	14948179	17p12	118220	Charcot-Marie-Tooth disease type 1A	PMP22
RP11-168B19	17	15067255	17p12	118220	Charcot-Marie-Tooth disease type 1A	PMP22
RP11-476N11	17	15185334	17p12	118220	Charcot-Marie-Tooth disease type 1A	PMP22
RP11-790K04	17	17157384	17p11.2	182290	Smith-Magenis syndrome	MED9
RP11-241J23	17	17326080.5	17p11.2	182290	Smith-Magenis syndrome	MED9
RP11-525O11	17	17565996	17p11.2	182290	Smith-Magenis syndrome	RAI1 SMCR5
RP1-253P7	17	17666333.5	17p11.2	182290	Smith-Magenis syndrome	RAI1 SMCR5
RP11-384M20	17	17836505.5	17p11.2	182290	Smith-Magenis syndrome	RAI1 SMCR5

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RP11-258F1	17	17952170	17p11.2	182290	Smith-Magenis syndrome	SMCR7 SMCR8
RP11-189D22	17	18027536	17p11.2	182290	Smith-Magenis syndrome	SMCR7 SMCR8
RP11-340I0	17	18229812	17p11.2	182290	Smith-Magenis syndrome	SMCR7 SMCR8
RP1-37N7	17	18262796	17p11.2	182290	Smith-Magenis syndrome	SMCR7 SMCR8
RP11-229K15	17	26373844	17q11.2	162200	Neurofibromatosis 1	NF1
RP11-353O18	17	26534994.5	17q11.2	162200	Neurofibromatosis 1	NF1
RP11-142O6	17	26574713	17q11.2	162200	Neurofibromatosis 1	NF1
RP1-41C23	17	26865303.5	17q11.2	162200	Neurofibromatosis 1	NF1
RP5-1037O7	17	67451553.5	17q24.3	114290	Campomelic dysplasia	SOX9
RP11-84E24	17	67600070	17q24.3	114290	Campomelic dysplasia	SOX9
RP11-95C11	17	67826017	17q24.3	114290	Campomelic dysplasia	SOX9
RP11-325E18	18	3412215.5	18p11.31	142946	Holoprosencephaly 4	TGIF
RP11-22C6	18	3525013.5	18p11.31	142946	Holoprosencephaly 4	TGIF
RP11-537E1	18	3539638	18p11.31	142946	Holoprosencephaly 4	TGIF
RP11-624I5	20	10484269.5	20p12.2	118450	Alagille syndrome	JAG1
RP5-1099D15	20	10600180	20p12.2	118450	Alagille syndrome	JAG1
RP11-2E8	20	10663129.5	20p12.2	118450	Alagille syndrome	JAG1
RP1-278O22	20	10683341.5	20p12.2	118450	Alagille syndrome	JAG1
RP11-567O17	20	10803268	20p12.2	118450	Alagille syndrome	JAG1
RP11-63O9	21	39363525.5	21q22.2	190685	Down syndrome critical region	DSCR2
RP11-367P1	21	39532816	21q22.2	190685	Down syndrome critical region	DSCR2
RP11-348C15	21	39603615.5	21q22.2	190685	Down syndrome critical region	DSCR2
RP11-466A11	21	44104900	21q22.3	236100	Holoprosencephaly 1	TMEM1
RP11-113I8	21	44255549	21q22.3	236100	Holoprosencephaly 1	TMEM1
RP11-317L14	21	44369577.5	21q22.3	236100	Holoprosencephaly 1	TMEM1
RP11-419G02	22	15885350	22q11.1	115470	Cat eye syndrome	CECR1 CECR5 CECR6
RP11-437O02	22	16013947	22q11.1	115470	Cat eye syndrome	CECR1 CECR5 CECR6
RP11-253P17	22	16033082.5	22q11.1	115470	Cat eye syndrome	CECR1 CECR5 CECR6
RP11-423L23	22	16042988.5	22q11.1	115470	Cat eye syndrome	CECR1 CECR5 CECR6
RP11-800B02	22	17559918.5	22q11.21	188400	DiGeorge syndrome	HIRA
RP11-243A04	22	17653998.5	22q11.21	188400	DiGeorge syndrome	HIRA
RP11-479G10	22	17724829.5	22q11.21	188400	DiGeorge syndrome	HIRA
RP11-488D20	22	17922281	22q11.21	188400	DiGeorge syndrome	HIRA
RP11-441P13	22	17922281	22q11.21	188400	DiGeorge syndrome	HIRA
RP11-652F11	22	18086824	22q11.21	192430	Velocardiofacial syndrome	TBX1
RP11-316L10	22	18179868	22q11.21	192430	Velocardiofacial syndrome	TBX1
RP11-330P17	22	18225548.5	22q11.21	192430	Velocardiofacial syndrome	TBX1
RP11-155B12	22	28308492	22q12.2	607379	Neurofibromatosis 2	NF2
RP11-772E21	22	28435913.5	22q12.2	607379	Neurofibromatosis 2	NF2
RP1-76B20	22	28465249	22q12.2	607379	Neurofibromatosis 2	NF2
RP11-423E19	22	49469303.5	22q13.33	606232	22q13.3 deletion syndrome	SHANK3
RP11-40G15	22	49486706	22q13.33	606232	22q13.3 deletion syndrome	SHANK3
RP11-109P4	X	5703160	Xp22.32	300495	Autism X-linked susceptibility to 2	NLGN4
RP11-769N24	X	5803056.5	Xp22.31	300495	Autism X-linked susceptibility to 2	NLGN4
RP11-594H22	X	5941386	Xp22.31	300495	Autism X-linked susceptibility to 2	NLGN4
RP11-323P4	X	6063815	Xp22.31	300495	Autism X-linked susceptibility to 2	NLGN4
RP11-44F2	X	6233748	Xp22.31	300495	Autism X-linked susceptibility to 2	NLGN4
RP11-483M24	X	7047424.5	Xp22.31	308100	Ichthyosis X-linked (steroid sulfatase deficiency)	STS
RP11-269D06	X	7247104.5	Xp22.31	308100	Ichthyosis X-linked (steroid sulfatase deficiency)	STS
RP13-302M16	X	7270373.5	Xp22.31	308100	Ichthyosis X-linked (steroid sulfatase deficiency)	STS
RP11-98J11	X	7299105.5	Xp22.31	308100	Ichthyosis X-linked (steroid sulfatase deficiency)	STS
RP11-323F16	X	7544773.5	Xp22.31	308100	Ichthyosis X-linked (steroid sulfatase deficiency)	STS
RP11-404I03	X	8605388	Xp22.31	308700	Kallmann syndrome 1	KAL1
RP11-163I1	X	11013218.5	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP11-425G22	X	11101225.5	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP1-27C22	X	11209882	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP11-450G14	X	11449560	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP11-361B16	X	11524624	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP11-403A11	X	11615027.5	Xp22.2	309801	Microphthalmia with linear skin defects	ARHGAP6
RP11-75D20	X	18320623	Xp22.13	312750	Rett syndrome	CDKL5
RP11-558P14	X	18363876.5	Xp22.13	312750	Rett syndrome	CDKL5
RP11-106N03	X	18456881.5	Xp22.13	312750	Rett syndrome	CDKL5
RP11-307L12	X	18620187.5	Xp22.13	312750	Rett syndrome	CDKL5
RP13-106B4	X	30040053.5	Xp21.2	300200	Adrenal Hypoplasia Congenital	MAGEB2 MAGEB4 NROB1
RP11-213O2	X	30104171	Xp21.2	300200	Adrenal Hypoplasia Congenital	MAGEB2 MAGEB4 NROB1
RP11-297K22	X	30116207.5	Xp21.2	300200	Adrenal Hypoplasia Congenital	MAGEB2 MAGEB4 NROB1
RP11-145B4	X	30337717.5	Xp21.2	307030	Hyperglycerolemia (glycerol kinase deficiency)	GK
RP11-420P1	X	30390650	Xp21.2	307030	Hyperglycerolemia (glycerol kinase deficiency)	GK
RP11-229K14	X	30550575	Xp21.2	307030	Hyperglycerolemia (glycerol kinase deficiency)	GK
RP11-242C19	X	30701881	Xp21.2	307030	Hyperglycerolemia (glycerol kinase deficiency)	GK
RP11-467P22	X	30779538.5	Xp21.2	307030	Hyperglycerolemia (glycerol kinase deficiency)	GK
RP11-340I10	X	30881676	Xp21.2	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-451N15	X	31001482	Xp21.2	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-346D13	X	31058679	Xp21.2	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-46A24	X	31101821	Xp21.2	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-609C15	X	31149398.5	Xp21.2	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-147B14	X	31353745.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD

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RP11-132A12	X	31397081	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-122N14	X	31508636	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-147E15	X	31598571.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-593P4	X	31625696	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-148I9	X	31800906.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-213G12	X	31842529	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-607K23	X	31862356	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-457L2	X	31959152	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-339E16	X	32001844	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-64I1	X	32158916	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP4-639D23	X	32210105.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP4-556A22	X	32314680	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP5-1147O16	X	32413262	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-142J18	X	32504457.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP1-37A12	X	32543302.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP5-1145L23	X	32695464.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP5-1174H9	X	32809822.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-151J4	X	32903177	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP5-1154A22	X	32998713.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP4-672M15	X	33074397	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP4-769D20	X	33185770	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP6-60B16	X	33322243.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-415K3	X	33408226.5	Xp21.1	310200	Muscular dystrophy (Duchenne Becker)	DMD
RP11-735G22	X	48287711	Xp11.23	190685	Down syndrome critical region	GATA1
RP11-695I24	X	48463763	Xp11.23	190685	Down syndrome critical region	GATA1
RP11-67K01	X	48510188.5	Xp11.23	190685	Down syndrome critical region	GATA1
RP11-297F13	X	48658849.5	Xp11.23	190685	Down syndrome critical region	GATA1
RP5-1091N2	X	70231468	Xq13.1	302800	Charcot-Marie-Tooth X-linked 1	GJB1
RP11-485H23	X	70347704.5	Xq13.1	302800	Charcot-Marie-Tooth X-linked 1	GJB1
RP11-554L02	X	70590990.5	Xq13.1	302800	Charcot-Marie-Tooth X-linked 1	GJB1
RP4-583H20	X	70619948	Xq13.1	302800	Charcot-Marie-Tooth X-linked 1	GJB1
RP11-402P6	X	70892957.5	Xq13.1	302800	Charcot-Marie-Tooth X-linked 1	GJB1
RP4-738A13	X	100463306	Xq22.1	300300	Bruton agammaglobulinemia tyrosine kinase	BTk
RP1-164F3	X	100568077.5	Xq22.1	300300	Bruton agammaglobulinemia tyrosine kinase	BTk
RP3-514P16	X	100638256.5	Xq22.1	300300	Bruton agammaglobulinemia tyrosine kinase	BTk
RP11-142J15	X	102860756.5	Xq22.2	312080	Pelizaeus-Merzbacher disease	PLP1
RP4-764D10	X	102905830.5	Xq22.2	312080	Pelizaeus-Merzbacher disease	PLP1
RP4-540A13	X	102969219.5	Xq22.2	312080	Pelizaeus-Merzbacher disease	PLP1
RP5-944N9	X	110320415	Xq23	300067	X-linked lissencephaly	DCX
RP5-914P14	X	110421070.5	Xq23	300067	X-linked lissencephaly	DCX
RP11-124N4	X	110572385.5	Xq23	300067	X-linked lissencephaly	DCX
RP6-170F5	X	110615291.5	Xq23	300067	X-linked lissencephaly	DCX
RP1-20J23	X	136340659	Xq26.3	306955	Heterotaxy, visceral, X-linked	ZIC3
RP11-81C12	X	136566124	Xq26.3	306955	Heterotaxy, visceral, X-linked	ZIC3
RP11-359I11	X	139275605	Xq27.1	300123	X-linked mental retardation with isolated growth hormone deficiency	SOX3
RP11-51C14	X	139381891	Xq27.1	300123	X-linked mental retardation with isolated growth hormone deficiency	SOX3
RP4-595A18	X	139501936	Xq27.1	300123	X-linked mental retardation with isolated growth hormone deficiency	SOX3
RP6-224C24	X	146593327	Xq27.3	309550	Fragile site mental retardation,1	FMR1
RP11-37P24	X	146829404	Xq27.3	309550	Fragile site mental retardation,1	FMR1
RP11-161L9	X	146886281	Xq27.3	309550	Fragile site mental retardation,1	FMR1
RP11-617G06	X	152850028	Xq28	312750	Rett syndrome	MECP2
RP4-671D9	X	152989984	Xq28	312750	Rett syndrome	MECP2
RP11-330B02	X	153109047	Xq28	312750	Rett syndrome	MECP2
RP11-400O10	Y	2745401.5	Yp11.31	480000	Sex-determining region Y	SRY
RP11-515L2	Y	2853630.5	Yp11.31	480000	Sex-determining region Y	SRY
RP11-414C23	Y	2931494	Yp11.31	480000	Sex-determining region Y	SRY
RP11-23N11	Y	3737486	Yp11.2	480000	Sex-determining region Y	SRY
RP11-125B02	Y	13349396	Yq11.21	415000	Azoospermia factor a (AZFa)	USP9Y
RP11-460B21	Y	13493611.5	Yq11.21	415000	Azoospermia factor a (AZFa)	USP9Y
RP11-414L16	Y	13690602.5	Yq11.21	415000	Azoospermia factor a (AZFa)	USP9Y
RP11-313L03	Y	13857049.5	Yq11.21	415000	Azoospermia factor a (AZFa)	UTY
RP11-218F6	Y	13884279.5	Yq11.21	415000	Azoospermia factor a (AZFa)	UTY
RP11-386L3	Y	14038175	Yq11.21	415000	Azoospermia factor a (AZFa)	UTY
RP11-509B6	Y	18563752	Yq11.221	415000	Azoospermia factor b (AZFb)	CDY2
RP11-157F24	Y	18942925.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-468D10	Y	18997045.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-243P9	Y	19184152	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-569J3	Y	19212392	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-143C1	Y	19377445.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-392F24	Y	19537559	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-207L19	Y	19716451	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-389F23	Y	19840369	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-118E9	Y	19948035.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-80E19	Y	20089071.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-169D1	Y	20127818	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2

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RP11-576C2	Y	20260574.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-424G14	Y	20311101.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-508P10	Y	20474272.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-356K22	Y	20570306	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-1285C3	Y	20723340.5	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-489O13	Y	20944145	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-329C15	Y	21073922	Yq11.222	415000	Azoospermia factor b (AZFb)	CDY2
RP11-256K9	Y	21194977	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-223K9	Y	21447148.5	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-339J4	Y	21554130	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-65G9	Y	21640012	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-470K20	Y	21763350	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-209I11	Y	21936117	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-220O2	Y	22089686.5	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2
RP11-178M5	Y	22230516	Yq11.223	415000	Azoospermia factor b (AZFb)	CDY2